From: Haviland, William A., Harald E.L. Prins, Dana Walrath and Bunny McBride. *Essence of Anthropology*. Cengage Learning; Boston, 2016.

From Zimmer, C. (2011). *Science ink: Tattoos of the science obsessed.*

VISUAL ESSENCE

In the 21st century, a biomedical doctor in Brazil might urge a patient to undergo testing for a specific disease gene as part of predicting the patient's future health. A pregnant woman in Canada might choose to terminate her pregnancy if prenatal testing reveals genes for a specific disease in a developing fetus. A pregnant woman in India, whose family considers female children a liability, could opt for prenatal genetic testing to ensure she will have a boy. Genetic analyses have become routine in biomedicine. The same is true for law and order. Throughout the globe, police identify criminals through DNA fingerprinting. They also maintain DNA databases of convicts and suspects for solving crimes in the future. In the United States, individuals wrongfully imprisoned for many years have been freed after genetic testing. No wonder, then, that many individuals have come to see genes and DNA as essential to individual identity. This correspondence between self and genetics has even prompted some people to tattoo an image of the genetic code into their very skin. But is the human condition this simple? Is DNA the essence of who we are? Do our genes determine our actions? And what will be the social consequences of depicting people as creatures programmed by their DNA, or by any other aspect of biology? A DNA tattoo, like the answers to these questions, embodies a constant interaction between human biology and culture.

Biology, Genetics, and Evolution

2

In this chapter you will learn to:

- **Compare evolution to creation stories.**
- **Identify the place of humans in the classification of all living things.**
- **Explain the molecular basis of evolution and the four evolutionary processes: mutation, gene flow, genetic drift, and adaptation.**
- **Describe how evolutionary processes account for the diversity of life on earth.**
- **Contrast how evolutionary processes work at the individual and population level.**
- **Explain how humans have adapted to their environments.**
- **Identify how new species come into being.**

Evolution and Creation Stories

The mythology of most peoples includes a story explaining the appearance of humans on earth. The Bible's Book of Genesis, for example, explains human origins. The Nez Perce, American Indians native to eastern Oregon and Idaho, provide us with a vastly different story. For the Nez Perce, human beings are the creation of Coyote, a trickster-transformer. Coyote chased the giant beaver monster Wishpoosh over the earth, leaving a trail to form the Columbia River. When Coyote caught Wishpoosh, he killed him, dragged his body to the riverbank, and cut it into pieces, each body part transforming into one of the various peoples of this region. The Nez Perce were made from Wishpoosh's head, thus conferring on them great intelligence and horsemanship (Clark, 1966).

Creation stories depict the relationship between humans and the rest of the natural world, sometimes reflecting a deep connection among people, other animals, and the earth. In the Nez Perce creation story, groups of people derive from specific body parts—each possessing a special talent and relationship with a particular animal. By contrast, the story of creation in Genesis—shared by Jews, Christians, and Muslims—emphasizes human uniqueness and the concept of time. Creation takes place as a series of actions over the course of six days. God's final act of creation is to fashion the first human in his own image before the seventh day of rest.

Like creation stories, evolution, the major organizing principle of the biological sciences, accounts for the diversity of life on earth. Evolution provides mechanisms for change and explanations for how the variety of organisms, both in the past and today, came into being. Evolution differs from creation stories in that it explains the diversity of life in a consistent scientific language, using testable ideas (hypotheses). Scientists make comparisons among living organisms to test hypotheses drawn from evolutionary theory. Scientists have deciphered the molecular basis of evolution and the mechanisms through which evolutionary forces work on populations of organisms. But scientific thought does not come out of a vacuum. As you will see, historical and cultural processes contribute to scientific thought.

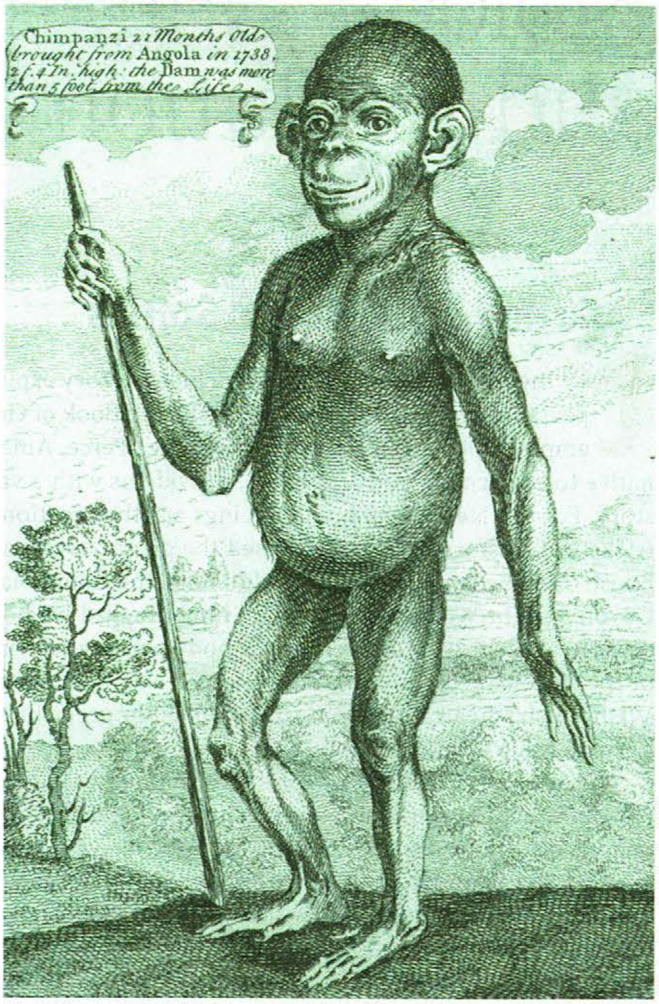
The Classification of Living Things

As European explorers exploited foreign lands, their approach to the natural world changed. The discovery of new life forms challenged the previously held notion of fixed, unchanging life on earth. The invention of instruments, such as the microscope to study the previously invisible interior of cells, also led to a new appreciation of life's diversity. Thus, Carolus Linnaeus (also known as Carl von Linné) developed the *Systema Naturae,* or system of nature, in the 18th century to classify the diversity of living things collected and brought to Europe by ship from throughout the globe. Linnaeus classified living things into a series of categories that are progressively more inclusive on the basis of internal and external visual similarities.

Noting the similarity among humans, monkeys, and apes, Linnaeus classified them together as **primates** (➤ **Figure 2.1**) one of several kinds of **mammals**, animals having body hair or fur who suckle or nurse their young. **Species**, the smallest working units in biological classificatory systems, are reproductively isolated populations or groups of populations capable of interbreeding to produce fertile offspring. Species are subdivisions of larger, more inclusive groups, called **genera** (singular, **genus**). Humans, for example, are classified in the genus *Homo* and the species *sapiens.*

Linnaeus based his classificatory system on the following criteria:

1. *Body structure:* A Guernsey cow and a Holstein cow are the same species because they have identical body structure. A cow and a horse do not.
2. *Body function:* Cows and horses give birth to live young. Although they are different species, they are closer than either cows or horses are to chickens, which lay eggs and have no mammary glands.
3. *Sequence of bodily growth:* At the time of birth—or hatching out of the egg—young cows and chickens possess body plans basically like that of their parents. They are therefore more closely related to each other than either one is to the frog, whose tadpoles undergo a series of changes before attaining the basic adult form.


Chimpanzi 21 Months Old brought from Angola in 1738. 2 f. 4 In. high: the Dam was more than 5 foot from the Life

© Hulton-Deutsch Collection/Corbis

➤ **Figure 2.1 Chimp or Human?** Early scientific struggles to classify great apes, and to identify and weigh the significance of the similarities and differences between them and humans, are reflected in early European renderings of apes, including this 18th-century image of a chimpanzee portrayed as a biped equipped with a walking stick.

Modern **taxonomy** (from the Greek for "naming divisions"), or the science of classification, retains the structure of the Linnaean system, but takes genetics as well as body structure, function, and growth into account to construct the relationships among living things. Molecular comparisons aimed at parasites, bacteria, and viruses allow scientists to trace the origins of particular diseases, such as swine flu, ebola, or HIV (human immunodeficiency virus). An emphasis on genetics has led to a reworking of taxonomic designation in the human family, among other families, as is described in ▶ **TABLE 2.1**.

Cross-species comparisons identify anatomical features of similar function as **analogies**, while anatomical features that have evolved from a common ancestral feature are called **homologies**. For example, the arm and hand of a human and the wing of a bat evolved from the front leg of a common ancestor. Although they have acquired

primates The group of mammals that includes lemurs, lorises, tarsiers, monkeys, apes, and humans.

mammals The class of vertebrate animals distinguished by bodies covered with hair or fur, self-regulating temperature, and in females, milk-producing mammary glands.

species The smallest working units in biological classificatory systems; reproductively isolated populations or groups of populations capable of interbreeding to produce fertile offspring.

genus (genera) In the system of plant and animal classification, a group of like species.

taxonomy The science of classification.

analogies In biology, structures possessed by different organisms that are superficially similar due to similar function but that do not share a common developmental pathway or structure.

homologies In biology, structures possessed by two different organisms that arise in similar fashion and pass through similar stages during embryonic development, although they may have different functions.

TABLE 2.1 | The Classification of Humans

Taxonomic Category	Category to Which Humans Belong	Biological Features Used to Define and Place Humans in This Category
Kingdom	Animalia	Humans are animals. We do not make our own food (as plants do) but depend upon intake of living food.
Phylum	Chordata	Humans are chordates. We have a notochord (a rodlike structure of cartilage) and nerve chord running along the back of the body as well as gill slits in the embryonic stage of our life cycle.
Subphylum*	Vertebrata	Humans are vertebrates, possessing an internal backbone with a segmented spinal column.
Class	Mammalia	Humans are mammals: warm-blooded animals covered with fur and possessing mammary glands for nourishing their young after birth.
Order	Primates	Humans are primates: a kind of mammal with a generalized anatomy, a relatively large brain, and grasping hands and feet.
Suborder	Anthropoidea	Humans are anthropoids: social, daylight-active primates.
Superfamily	Hominoid	Humans are hominoids with broad, flexible shoulders and no tail. Chimps, bonobos, gorillas, orangutans, gibbons, and siamangs are also hominoids.
Family/Subfamily	Hominid/Hominin	Humans are hominids. We are hominoids from Africa, genetically more closely related to chimps, bonobos, and gorillas than to hominoids from Asia. Some scientists use "hominid" to refer only to humans and their ancestors. Others include chimps and gorillas in this category, using the subfamily "hominin" to distinguish humans and their ancestors from chimps and gorillas and their ancestors. Hominin emphasizes the close genetic relationship among humans, chimps, and gorillas. Hominid gives preference to the similarities in body shape among chimpanzees, gorillas, and orangutans.
Genus Species	*Homo sapiens*	Humans have large brains and rely on cultural adaptations to survive. Genus and species names are always italicized.

*Most categories can be expanded or narrowed by adding the prefix "sub" or "super." A family could thus be part of a superfamily and in turn contain two or more subfamilies.

different functions, they are homologous structures (➤ **Figure 2.2**). During their early embryonic development, homologous structures arise in a similar fashion and pass through similar stages before differentiating. The wings of birds and butterflies look similar and have a similar function (flying): These are analogous structures because they do not follow the same developmental sequence.

When constructing evolutionary relationships, only homologies matter. Careful comparison and analysis have allowed scientists to group species into genera and into even larger groups such as families, orders, classes, phyla, and kingdoms. Characteristics shared by all the organisms in the group define each taxonomic level.

The Discovery of Evolution

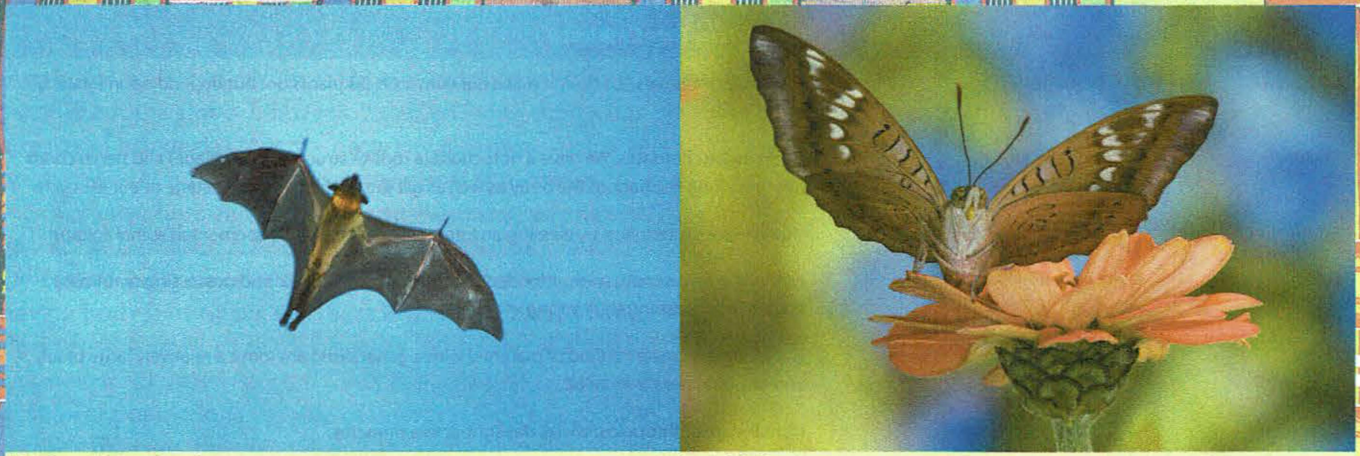
Just as European seafaring and exploitation brought about an awareness of the diversity of life across the globe, industrialization in Europe brought about an awareness of change in life forms through time. As workers cut away the earth to lay railway tracks and excavated limestone for buildings, fossils were brought into the light. These discoveries revolutionized scientific thought.

At first, religious doctrine was used to interpret fossilized remains of elephants and giant saber-toothed tigers in Europe. The early 19th-century theory of *catastrophism*, championed by French paleontologist George Cuvier, invoked the great flood described in Genesis to account for the disappearance of these species on European lands.

Another French scientist, Jean-Baptiste Lamarck, was among the first to suggest a mechanism to account for life's diversity that did not rely upon scriptures. His theory of the *inheritance of acquired characteristics* proposed that behavior brought about changes in organisms' forms, such as the famed first giraffe who gained its long neck by stretching to reach leaves on the highest treetop branches and passed this acquired trait onto its offspring. Lamarck was the first to connect organisms with the environments they inhabit, and the mechanism he proposed for change over time works perfectly for qualities inherited via culture. Recent studies on how genes get turned on and off also demonstrate a place for Lamarckian biological inheritance.

At about the same time that Lamarck formulated his ideas, British geologist Sir Charles Lyell proposed a nonreligious theory to account for variations in the earth's surface. His theory, *uniformitarianism*, maintained that

VISUAL COUNTERPOINT



Mark Carwardine/Photolibrary/Getty Images

Darrell Gulin/Getty Images

➤ **Figure 2.2 Homology and Analogy** The bat wing is homologous to the human hand. Look closely at the bones supporting the wing, and you can see that they are the same bones found in the human arm and hand. Homologous structures have the same embryonic origins but ultimately take on different functions. For humans, our grasping hands function in a variety of ways, including even language. Bats use these same bones for flight. By contrast, the wing of a butterfly, though used for flight, is analogous to the bat wing. These wings have similar functions though they arise from completely different structures.

immediately observable changes in the earth's surface from erosion and other natural processes could account for variation in the earth's surface if they took place over extremely long periods of time. Lyell's theory was incompatible with religious doctrine because the length of time required for uniformitarianism far exceeded the 6,000-year age of the earth contained in the Bible. By the start of the 19th century, many naturalists had come to accept the idea that the earth was old and that life had evolved, even though they were not clear about how it happened. It remained for Charles Darwin (1809–1882) to formulate a theory that has withstood the test of time.

Charles Darwin began the study of medicine at the University of Edinburgh in Scotland. Finding himself unfit for this profession, he went to Christ's College, Cambridge, to study theology. He then left Cambridge to take the position of companion to Captain Robert FitzRoy aboard the HMS *Beagle*, embarking on an expedition to various poorly mapped parts of the world. The voyage lasted almost five years, taking Darwin along the coasts of South America, to the Galapagos Islands, across the Pacific to Australia, and then across the Indian and Atlantic oceans to South America before returning to England in 1836 (➤ **Figure 2.3**).

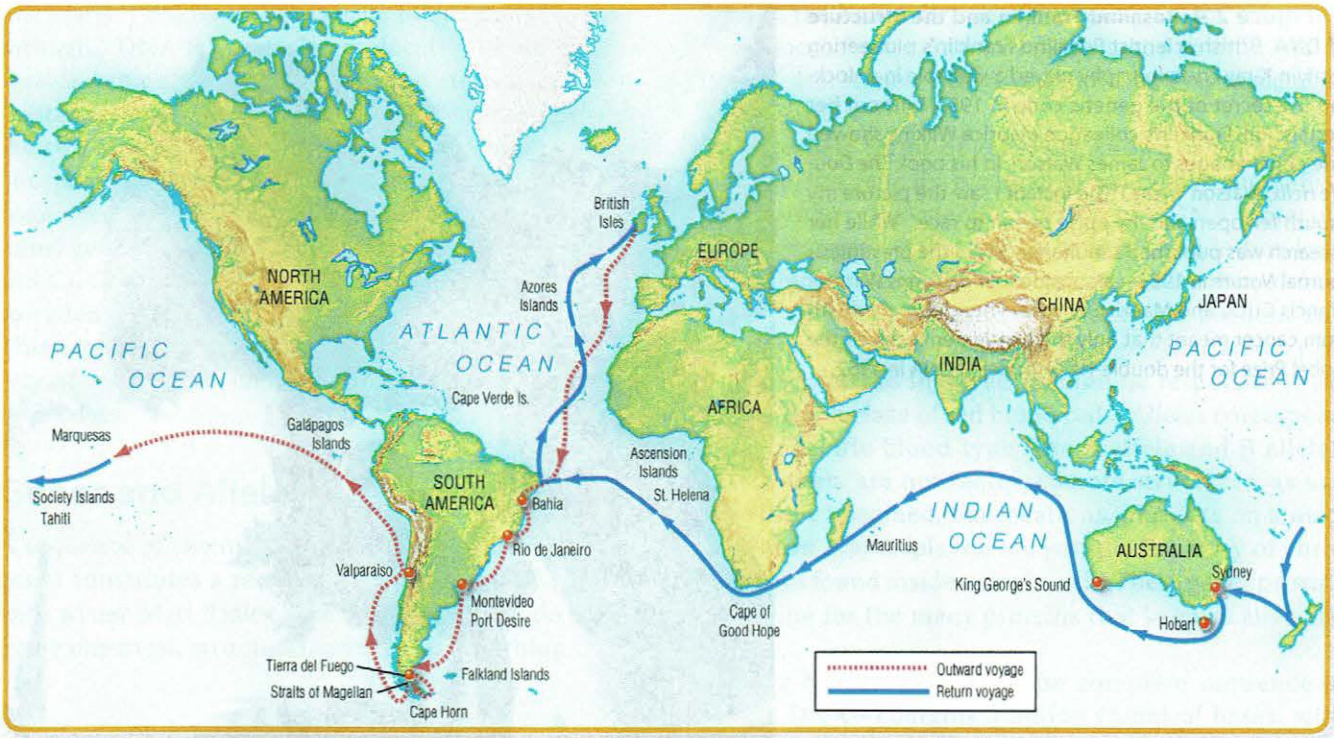
Observing the tremendous diversity of living creatures as well as the fossils of extinct animals, Darwin noted that species varied according to the environments they inhabited. The observations he made on this voyage, his readings of Lyell's geological theories, and arguments he had with Captain FitzRoy all contributed to Darwin's most famous book, *On the Origin of Species*. Published in 1859, twenty years after he returned from his voyage, this book accounts for change within species and for the emergence of new species in purely naturalistic terms.

Darwin added observations from English farm life and intellectual thought to his observations while on the *Beagle*. He paid particular attention to domesticated animals and farmers' "artificial selection," a practice of breeding stock to select for specific traits. Darwin's theoretical breakthrough derived from an essay by economist Thomas Malthus (1766–1834), which warned of the potential consequences of increased human population, particularly of the poor. Malthus observed that animal populations, unlike human populations, remained stable, due to an overproduction of young followed by a large proportion of animal offspring not surviving to maturity.

Darwin combined his observations into the theory of **natural selection** as follows: All species display a range of

natural selection The evolutionary process through which factors in the environment exert pressure, favoring some individuals over others to produce the next generation.

From Jurmain, R., Kilgore, L., Trevathan, W., & Ciochon, R. (2011–2012). *Introduction to physical anthropology* (13th ed.).
© 2012 Cengage Learning Inc.


British Isles
EUROPE
NORTH AMERICA
Azores Islands
ATLANTIC OCEAN
CHINA
JAPAN
PACIFIC OCEAN
PACIFIC OCEAN
INDIA
Cape Verde Is.
AFRICA
Marquesas
Galápagos Islands
SOUTH AMERICA
Ascension Islands
St. Helena
Bahia
Society Islands
Tahiti
INDIAN OCEAN
Rio de Janeiro
Mauritius
AUSTRALIA
Valparaiso
Montevideo
Port Desire
King George's Sound
Sydney
Cape of Good Hope
Hobart
Tierra del Fuego
Straits of Magellan
Falkland Islands
Cape Horn
Outward voyage
Return voyage

➤ **Figure 2.3** **The Voyage of the HMS *Beagle*** During his journey of almost five years, Darwin was exposed to the diversity of plant and animal life, as well as to the varied environments across the globe. This planted the seeds of his evolutionary theory.

variation, and all have the ability to expand beyond their means of subsistence. It follows that, in their "struggle for existence," organisms with variations that help them to survive in particular environments will reproduce with greater success than those without such variations. Thus, over time, nature selects the most advantageous variations and species evolve. In retrospect, the idea seemed so obvious that Thomas Henry Huxley, one of the era's most prominent scientists, remarked, "How extremely stupid of me not to have thought of that" (quoted in Durant, 2000, p. 11).

However straightforward the idea of evolution by natural selection may appear, the theory was (and continues to be) a source of considerable controversy. Two problems plagued Darwin's theory throughout his career: First, how did variation initially arise? Second, what was the mechanism of heredity by which traits could be passed from one generation to the next?

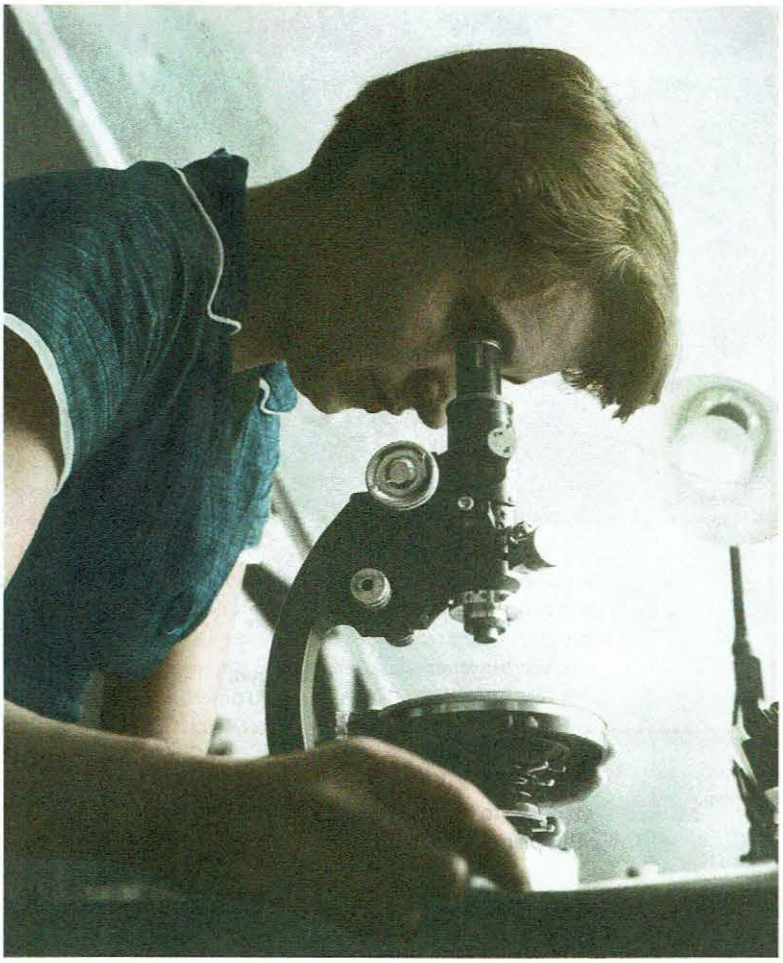
Heredity

Ironically, some of the information Darwin needed was available by 1866. Gregor Mendel (1822–1884), a Roman Catholic monk, developed the basic laws of heredity while working in the monastery gardens in Brno, a city in today's Czech Republic. Mendel, who was raised on a farm, possessed two particular talents: a flair for mathematics and a passion for gardening. As with all farmers of his time, Mendel had an intuitive understanding of biological inheritance. He went a step further, though, in that he recognized the need for theoretical explanations. At age 34, he began careful breeding experiments in the monastery garden, starting with pea plants.

Over eight years, Mendel planted more than 30,000 plants, controlling their pollination, observing the results, and figuring out the mathematics behind it all. This allowed him to unravel the basic laws of heredity. Though his findings were published in 1866 in a respected scientific journal, no one recognized the importance of Mendel's work during his lifetime.

In 1900, cell biology had advanced to the point where rediscovery of Mendel's laws was inevitable, and in that year three European botanists, working independently of one another, rediscovered not only the laws but also Mendel's original paper. With this rediscovery, the science of genetics began. Still, it would be another fifty-three years before the molecular mechanisms of heredity and the discrete units of inheritance would be discovered. Today, a comprehensive understanding of heredity, molecular genetics, and population genetics supports Darwinian evolutionary theory.

➤ Figure 2.4 Rosalind Franklin and the Structure of DNA British scientist Rosalind Franklin's pioneering work in X-ray crystalography played a vital role in unlocking the secret of the genetic code in 1953. Without her permission, Franklin's colleague Maurice Wilkins showed one of her images to James Watson. In his book *The Double Helix,* Watson wrote, "The instant I saw the picture my mouth fell open and my pulse began to race." While her research was published simultaneously in the prestigious journal *Nature* in 1953—alongside that of James Watson, Francis Crick, and Maurice Wilkins—her untimely death from cancer meant that only the gentlemen received the Nobel Prize for the double-helix model of DNA in 1962.



Science Source

The Transmission of Genes

We define **genes** as portions of the DNA molecule containing a sequence of base pairs that encodes a particular protein. But when biologists coined the term from the Greek word for "birth" at the turn of the 20th century, the molecular basis of the gene was still fifty years away from discovery. Mendel had deduced the presence and activity of genes by experimenting with garden peas to determine how various traits are passed from one generation to the next. Specifically, he discovered that inheritance was *particulate,* rather than *blending.* The units controlling the expression of visible traits come in pairs, one from each parent, and retain their separate identities over the generations rather than blending into a combination of parental traits in offspring. This was the basis of Mendel's first **law of segregation**, which states that pairs of genes separate, keep their individuality, and are passed on to the next generation unaltered. Another finding—Mendel's **law of independent assortment**—states that different traits (under the control of distinct genes) are inherited independently of one another.

Mendel based his laws on statistical frequencies of observed characteristics, such as color and texture in generations of plants. When **chromosomes**, the cellular structures containing the genetic information, were discovered at the start of the 20th century, they provided a visible vehicle for transmission of traits proposed in Mendel's laws.

Then in 1953, James Watson and Francis Crick discovered the mechanism for inheritance based on the structure of **DNA (deoxyribonucleic acid)**—long strands that form chromosomes. (Rosalind Franklin, shown in ➤ **Figure 2.4**,

genes The portions of DNA molecules that direct the synthesis of specific proteins.

law of segregation The Mendelian principle that variants of genes for a particular trait retain their separate identities through the generations.

law of independent assortment The Mendelian principle that genes controlling different traits are inherited independently of one another.

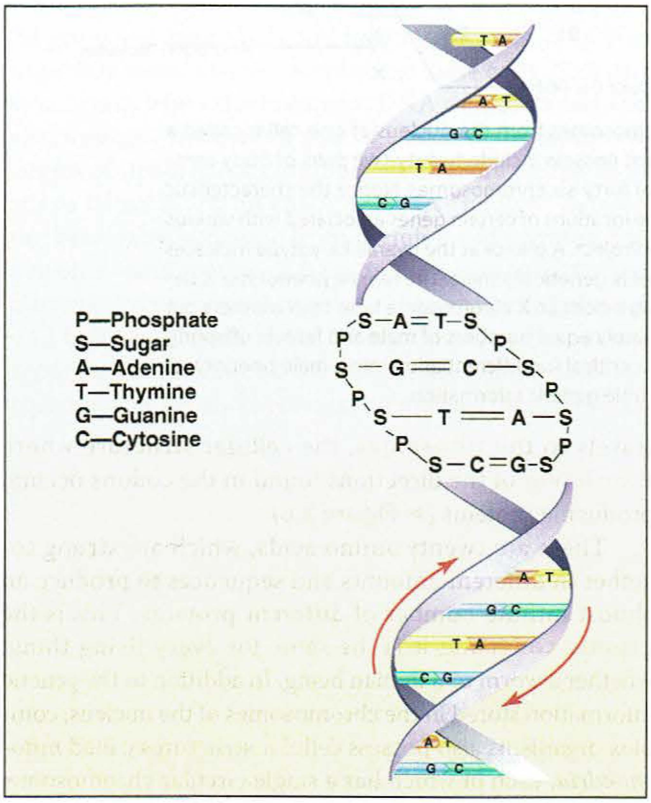
chromosomes In the cell nucleus, the structures visible during cellular division containing long strands of DNA combined with a protein.

DNA (deoxyribonucleic acid) The genetic material consisting of a complex molecule whose base structure directs the synthesis of proteins.

was a largely unknown contributor to this amazing breakthrough.) DNA is a complex molecule with an unusual *double-helix* shape, rather like two strands of a rope twisted around each other with ladderlike steps between the two strands (➤ **Figure 2.5**). Alternating sugar and phosphate molecules form the backbone of these strands connected to each other by four base pairs: adenine, thymine, guanine, and cytosine (usually written as A, T, G, and C). Connections between the strands occur between so-called complementary pairs of bases (A to T, G to C). This arrangement confers upon genes the unique property of replication—being able to make exact copies of themselves.

Genes and Alleles

A sequence of chemical bases on a molecule of DNA (a gene) constitutes a recipe for making proteins. As science writer Matt Ridley puts it, "Proteins . . . do almost every chemical, structural, and regulatory thing that is done in the body: they generate energy, fight infection, digest food, form hair, carry oxygen, and so on" (Ridley, 1999, p. 40). Almost everything in the body is made of or by proteins.



➤ **Figure 2.5** **The Structure of DNA** This diagrammatic representation of a portion of DNA (deoxyribonucleic acid) illustrates its twisted ladderlike structure. Alternating sugar and phosphate groups form the structural sides of the ladder. The connecting "rungs" are formed by pairings between complementary bases—adenine with thymine and cytosine with guanine.

Alternate forms of genes are known as **alleles**. For example, the gene for a human blood type in the A-B-O system refers to a specific portion of a DNA molecule on chromosome 9 that in this case is 1,062 letters long (a medium-sized gene). This gene specifies the production of an **enzyme**, a kind of protein that initiates and directs a chemical reaction. This particular enzyme causes molecules involved in immune responses to attach to the surface of red blood cells. Alleles correspond to the specific blood type (the A allele and B allele). Genes, then, are not really separate structures, as had once been imagined, but locations, like dots on a map. (➤ **Figure 2.6**) displays a **karyotype**, the array of chromosomes found inside a single cell.) These genes provide the recipe for the many proteins that keep us alive and healthy.

The human **genome**—the complete sequence of human DNA—contains 3 billion chemical bases, with 20,000 to 25,000 genes, a number similar to that found in most mammals. Of the 3 billion bases, humans and mice are about 90 percent identical. Both species have three times as many genes as does the fruit fly, but surprisingly humans and mice have half the number of genes found in the rice plant! At the same time, those 20,000 to 25,000 human genes account for only 1 to 1.5 percent of the entire genome, indicating that scientists still have far more to learn about how genes work. Frequently, genes themselves are split by long stretches of DNA that are not part of the known protein code; for example, the 1,062 bases of the A-B-O blood-group gene are interrupted by five such stretches. In the course of protein production, these stretches of DNA are metaphorically snipped out and left on the cutting room floor.

How is the DNA recipe converted into a protein? Through a series of intervening steps, each three-base sequence of a gene, called a **codon**, specifies production of a particular amino acid, strings of which build proteins.

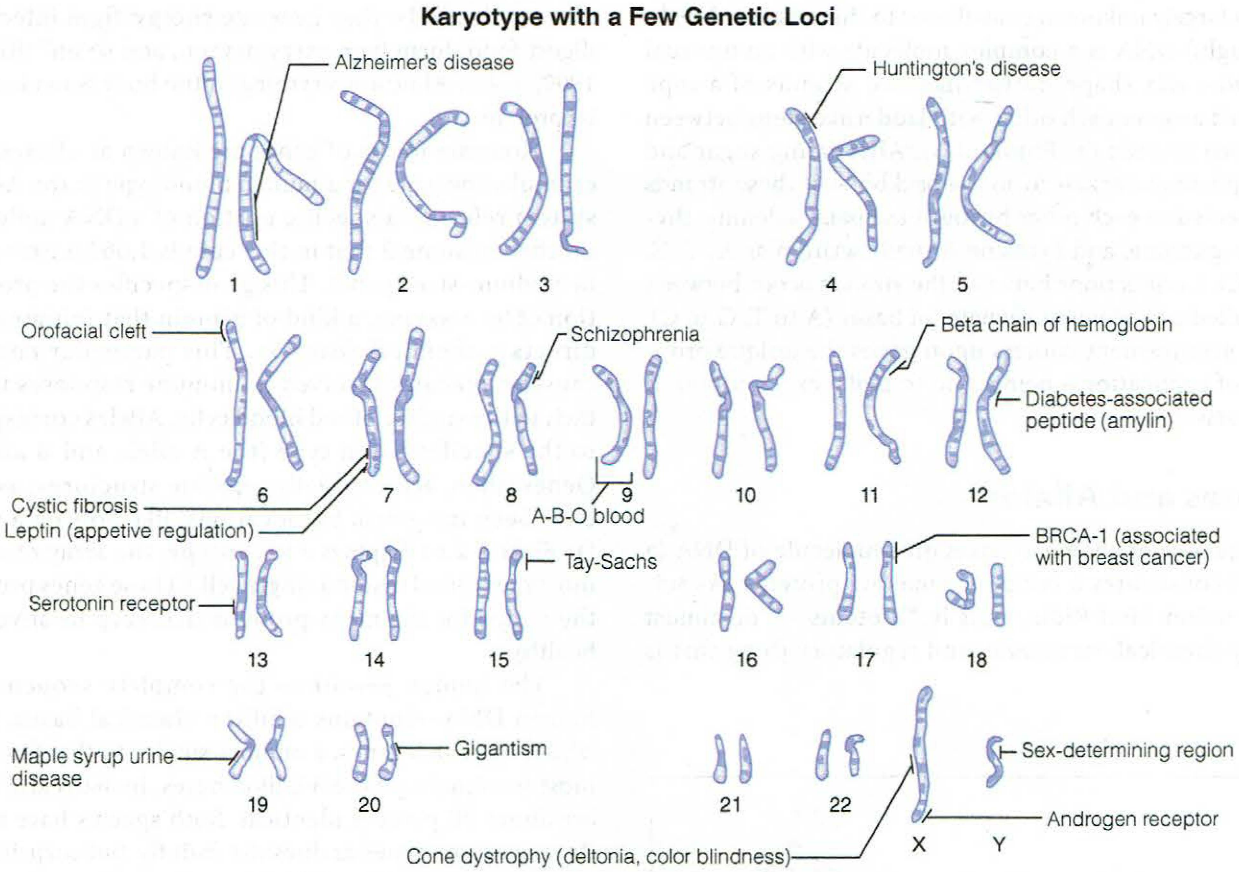
alleles Alternate forms of a single gene.

enzymes Proteins that initiate and direct chemical reactions.

karyotype The array of chromosomes found inside a single cell.

genome The complete structure sequence of DNA for a species.

codon Three-base sequence of a gene that specifies a particular amino acid for inclusion in a protein.


Karyotype with a Few Genetic Loci
Alzheimer's disease
Huntington's disease
1
2
3
4
5
Orofacial cleft
Schizophrenia
Beta chain of hemoglobin
Diabetes-associated peptide (amylin)
6
7
8
9
10
11
12
Cystic fibrosis
Leptin (appetive regulation)
A-B-O blood
Tay-Sachs
BRCA-1 (associated with breast cancer)
Serotonin receptor
13
14
15
16
17
18
Maple syrup urine disease
Gigantism
Sex-determining region
19
20
21
22
Androgen receptor
X
Y
Cone dystrophy (deltonia, color blindness)

➤ Figure 2.6 **A Human Karyotype** An array of chromosomes from the nucleus of one cell is called a karyotype. The twenty-three pairs of chromosomes humans possess include twenty-two pairs of body chromosomes, plus one pair of sex chromosomes, for a total of forty-six chromosomes. Notice the characteristic shape and relative size of each of the chromosomes and the locations of certain genes associated with various diseases and conditions identified by the Human Genome Project. A glance at the overall karyotype indicates a normal number of chromosomes and that this individual is genetically male. The female phenotype is determined by the presence of two X chromosomes. Offspring inherit an X chromosome from their mothers but either an X or a Y from their fathers, resulting in approximately equal numbers of male and female offspring in subsequent generations. Although the Y chromosome is critical for differentiation into a male phenotype, compared to other chromosomes, the Y is tiny and carries little genetic information.

Because DNA cannot leave the cell's nucleus (**➤ Figure 2.7**), the directions for a specific protein are first converted into **RNA (ribonucleic acid)** in a process called **transcription**. RNA is single stranded and differs from DNA in the structure of its sugar-phosphate backbone and in the presence of the base uracil (U) rather than thymine. Next, the RNA travels to the **ribosomes**, the cellular structure where **translation** of the directions found in the codons occurs, producing proteins (**➤ Figure 2.8**).

RNA (ribonucleic acid) Similar to DNA but with uracil substituted for the base thymine. Transcribes and carries instructions from DNA from the nucleus to the ribosomes, where it directs protein synthesis. Some simple life forms contain RNA only.

transcription The process of conversion of instructions from DNA into RNA.

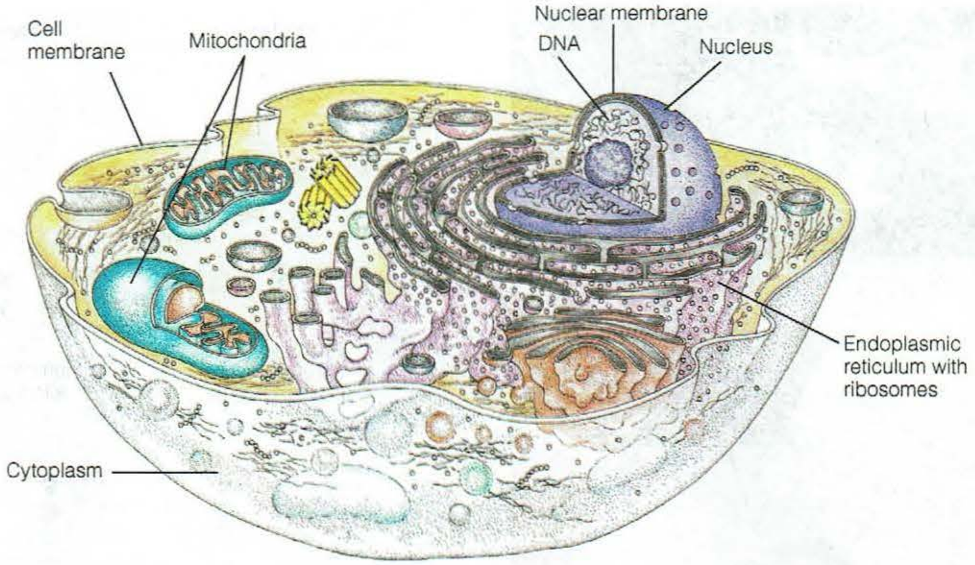
ribosomes Structures in the cell where translation occurs.

translation In genetics, the process of conversion of RNA instructions into proteins.

genetic code The sequence of three bases (a codon) that specifies the sequence of amino acids in protein synthesis.

There are twenty amino acids, which are strung together in different amounts and sequences to produce an almost infinite number of different proteins. This is the **genetic code**, and it is the same for every living thing, whether a worm or a human being. In addition to the genetic information stored in the chromosomes of the nucleus, complex organisms also possess cellular structures called *mitochondria,* each of which has a single circular chromosome. On the other end of the spectrum, simple living things without nucleated cells, such as the retrovirus that causes AIDS, contain their genetic information only as RNA.

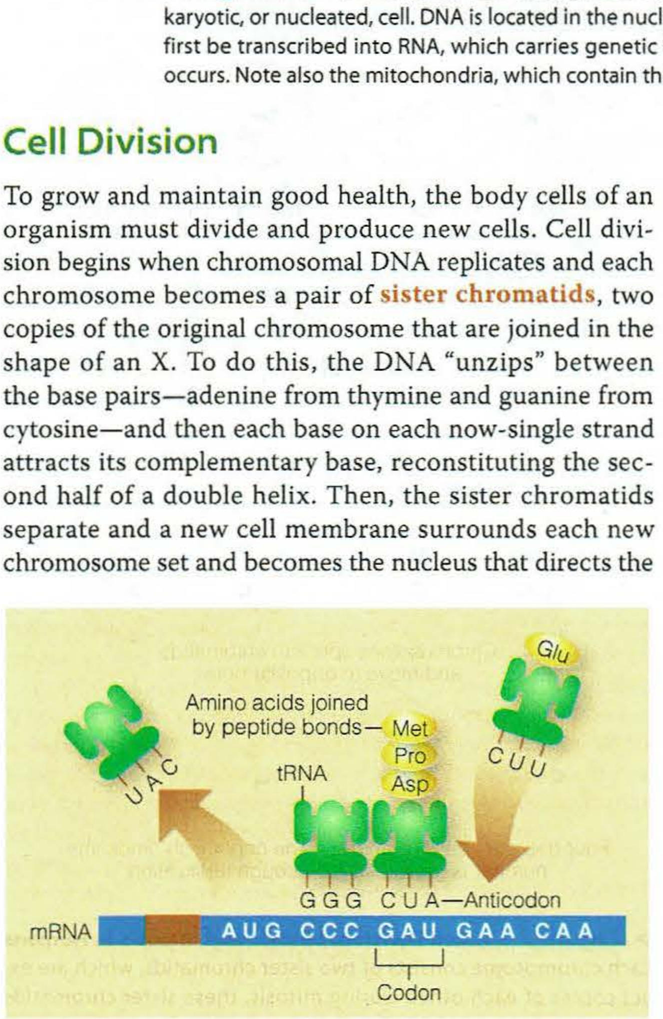
Mistakes can occur in the replication process, adding or subtracting repeats of the four bases: A, C, G, and T. This happens with some frequency and differently in every individual. As these "mistakes" accumulate over time, people develop their unique DNA fingerprint.



➤ **Figure 2.7** **A Eukaryotic Cell** The figure shows the three-dimensional structure of a generalized eukaryotic, or nucleated, cell. DNA is located in the nucleus. Because DNA cannot leave the nucleus, genes must first be transcribed into RNA, which carries genetic information to the ribosomes, where protein synthesis occurs. Note also the mitochondria, which contain their own circular chromosomes and mitochondrial DNA.

Cell Division

To grow and maintain good health, the body cells of an organism must divide and produce new cells. Cell division begins when chromosomal DNA replicates and each chromosome becomes a pair of **sister chromatids**, two copies of the original chromosome that are joined in the shape of an X. To do this, the DNA "unzips" between the base pairs—adenine from thymine and guanine from cytosine—and then each base on each now-single strand attracts its complementary base, reconstituting the second half of a double helix. Then, the sister chromatids separate and a new cell membrane surrounds each new chromosome set and becomes the nucleus that directs the activities of a new cell. This kind of cell division is called **mitosis**. Barring errors in this replication process, cells divide mitotically to form daughter cells that are exact genetic copies of the parent cell.



➤ **Figure 2.8** **From DNA into Proteins** Codons of DNA (a sequence of three bases) are transcribed into the complementary codons of RNA to leave the nucleus. In the ribosomes, these codons are translated into proteins as amino acids are strung together into the specified chains. Can you think of the bases that would have been found in the DNA that correspond to the section of RNA pictured here?

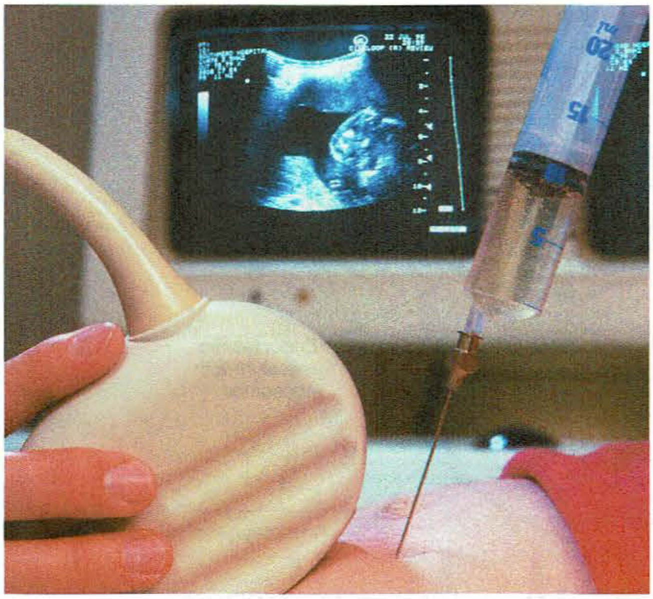
Like most animals, humans reproduce sexually. The "popularity" of sex from an evolutionary perspective derives from the genetic variation that it provides. All animals contain two versions of each chromosome, having inherited one from each parent. In humans this involves twenty-three pairs of chromosomes. Sexual reproduction can bring favorable alleles together, purge the genome of harmful ones, and allow beneficial alleles to spread without being held back by the baggage of disadvantageous variants of other genes.

Although human societies have always regulated sexual reproduction in some ways, the science of genetics has had a tremendous impact on social aspects of reproduction. Among wealthier parents-to-be, prenatal genetic testing has become increasingly common. As mentioned in the opening of this chapter, such testing aims to eliminate conditions not favored within a society. For example, in India, where female children are undervalued, prenatal genetic testing has led to selective abortion of female fetuses (➤ **Figure 2.9**) (Arnold, Kishor, & Roy, 2002).

No aspect of human reproduction or genetics is simply biological. Social and political processes impact the interpretation and use of genetic technology. The technology,

sister chromatid One half of the X shape of a chromosome visible after replication. Each half is a copy of the original chromosome.

mitosis A kind of cell division that produces new cells having exactly the same number of chromosome pairs, and hence copies of genes, as the parent cell.



Saturn Stills/Science Source

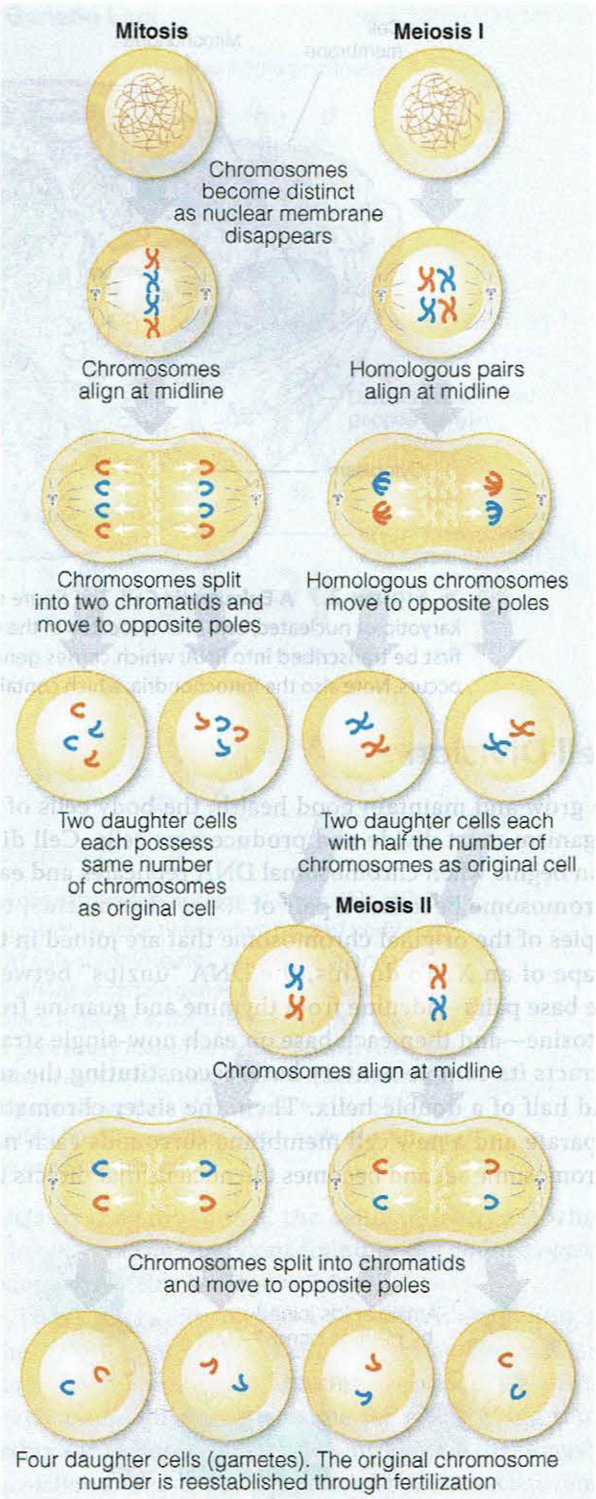
➤ **Figure 2.9** **Prenatal Genetic Testing by Amniocentesis** Prenatal genetic testing is conducted most frequently by amniocentesis, a technique that began in the 1960s through which a medical practitioner draws fluid containing cells from a developing embryo from the womb of a pregnant woman. Lab technicians then analyze the chromosomes and specific genes for abnormalities. Cultural anthropologists have shown that a biological fact (such as an extra twenty-first chromosome or Down syndrome) is open to diverse interpretations and reproductive choices by "potential parents" (Rapp, 1999). New reproductive technologies have far-reaching social consequences. Genetic testing may lead to the labeling of certain people as undesirable, pitting women's reproductive rights against the rights of the disabled.

in turn, shapes social definitions of family, identity, and the types of citizens preferred by a given society. This chapter's Biocultural Connection tells of how DNA testing has made its way into the lives of African refugees seeking reunification with their families in the United States.

Sexual reproduction increases genetic diversity, which in turn contributes to adaptation among sexually reproducing species. Sexual reproduction involves the merging of two cells, one from each parent, to make a new individual. If two regular body cells, each containing twenty-three pairs of chromosomes, were to merge, the lethal result would be a new individual with forty-six pairs of chromosomes. Instead, sexual reproduction involves joining specialized sex cells (eggs and sperm) produced by a different kind of cell division, called **meiosis**.

Meiosis begins like mitosis, with the replication and doubling of the original chromosomes through the formation of sister chromatids. Cells then proceed to divide into four new cells, each of which has half the number of chromosomes compared to the parent cell (➤ **Figure 2.10**).

meiosis A kind of cell division that produces the sex cells, each of which has half the number of chromosomes found in other cells of the organism.


Mitosis
Meiosis I
Chromosomes become distinct as nuclear membrane disappears
Chromosomes align at midline
Homologous pairs align at midline
Chromosomes split into two chromatids and move to opposite poles
Homologous chromosomes move to opposite poles
Two daughter cells each possess same number of chromosomes as original cell
Two daughter cells each with half the number of chromosomes as original cell
Meiosis II
Chromosomes align at midline
Chromosomes split into chromatids and move to opposite poles
Four daughter cells (gametes). The original chromosome number is reestablished through fertilization

➤ **Figure 2.10** **Cell Division: Mitosis and Meiosis in Humans** Each chromosome consists of two sister chromatids, which are exact copies of each other. During mitosis, these sister chromatids separate into two identical daughter cells. In meiosis, the cell division responsible for the formation of gametes, the first division halves the chromosome number. The second meiotic division is essentially like mitosis and involves the separation of sister chromatids. Chromosomes in red came from one parent; those in blue came from the other. Meiosis results in four daughter cells that are not identical.

BIOCULTURAL CONNECTION

Bonds beyond Blood: DNA Testing and Refugee Family Unification

By Jason Silverstein

In February 2008, the U.S. government began to assess the use of DNA testing as documentary proof of familial relationship among asylum seekers from Africa attempting reunification with family members already in the United States. The pilot study began with 500 residents of the Dadaab refugee camp in Nairobi, Kenya, where an estimated 465,000 refugees currently live in a space designed for 90,000. DNA testing was later expanded to include 3,000 refugees in Ethiopia, Uganda, Ghana, Guinea, and Côte d'Ivoire.

The pilot program operated on the assumption of "guilty until proven family." Anything other than DNA proof of relationship was recorded as fraud. Refusal to test was recorded as fraud. On a petition with multiple family members, if one person refused, did not show up for, or failed the test, then the entire petition was recorded as fraud. Shockingly, the "anchor" (the person with whom the applicants desired to reunite) was never tested. Only the relationships between individuals on the application were tested. By the time the pilot phase ended, these policies resulted in the classification of 80 percent of family reunification claims as fraudulent, and the U.S. government suspended the reunification program. As DNA testing increasingly becomes the standard by which border security officials investigate kinship claims, what makes a family legitimate will be determined by the social prescriptions of the receiving community.

Reuters/Jonathan Ernst/Landov

Covering 50 square kilometers in northeastern Kenya, the Dadaab refugee camp is the largest in the world. Each day, over 1,300 new refugees arrive seeking sanctuary from political persecution and starvation brought about by civil war and drought in their homelands. Here, Somali refugees pray during Eid al-Fitr, marking the end of the Muslim holy month of Ramadan.

The definition of *family* is not necessarily portable across borders. Refugees are forced to conform their qualifications of family and, thus, their very life stories to the social norms of the receiving community. We should not confuse the neutrality of DNA as hereditary material with the neutrality of those who collect, process, and interpret it. Far from value-neutral technology, DNA testing for family reunification reveals an allegiance to a particular social universe and often conceals the reality of the refugees' lived experience.

One especially lucid example of this claim is provided by a refugee case manager who remarked that polygamous families (those with more than one spouse) are never resettled (in other words, would never pass the family-relatedness test). Given the testing protocol of the pilot project (testing the genetic relationship between applicants and not between the applicant and the anchor), one readily can imagine that a mother as a primary applicant may not share DNA with her child.

Family is not simply genetic or socially prescribed; it is also existentially evolved, especially for refugees who have spent years if not decades in camps. An interviewer with the United Nations High Commissioner for Refugees related stories about parents who did not want to distinguish between their biological and adopted, often war-orphaned, children. Given that the verification interview takes place in front of the children, avoiding this distinction may have little to do with what security officials cruelly call fraud or abuse. As one case manager starkly put it, the DNA testing methodology overlooks that those who survive often do not survive unscathed.

Biocultural Questions

When DNA testing is used by authorities for identification purposes, what rifts open between the truth of one's life and what can be extracted from one's blood? What should we be searching for when we want to know who a person truly is?

Adapted from Silverstein, J. (2012). Bonds beyond blood: DNA testing and refugee family resettlement. *Anthropology News 53* (4), 11.

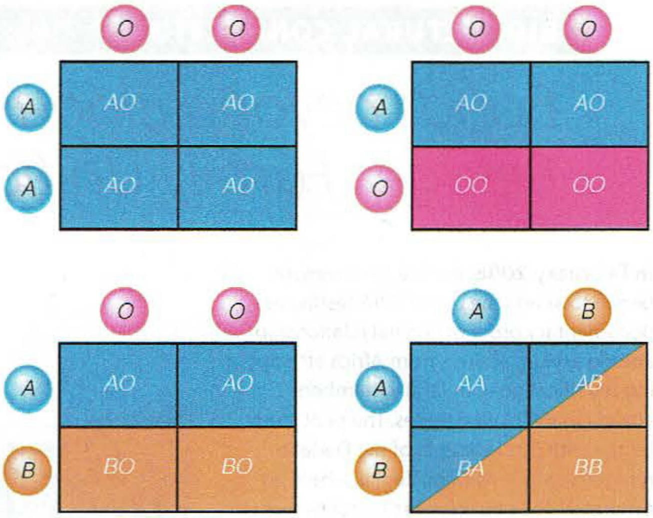
Human eggs and sperm have only twenty-three single chromosomes (half of a pair), whereas body cells have twenty-three pairs, or forty-six chromosomes.

Meiosis has important implications for genetics. Because paired chromosomes are separated, the daughter cells will not be identical. Two of the four new cells will have half of each pair of chromosomes, and the other two will have the second half of the original chromosome pair. In addition, corresponding portions of one chromosome may "cross over" to the other one, somewhat scrambling the genetic material compared to the original chromosomes.

Sometimes, the original pair is **homozygous**, possessing identical alleles for a specific gene. For example, if both chromosomes of the original pair the gene for A-B-O blood type possess the allele for type A blood, then all new cells will have the A allele. But if the original pair is **heterozygous**, with the A allele on one chromosome and the B allele on the other, then half of the new cells will contain only the B allele; the offspring have a 50–50 chance of getting either one.

What happens when a child inherits the allele for type O blood from one parent and that for type A from the other? Will the child have blood of type A, O, or some mixture of the two? ➤ **Figure 2.11** illustrates some of the possible outcomes. Many of these questions were answered by Mendel's original experiments.

Mendel discovered that certain alleles can mask the presence of others; one allele is **dominant**, whereas the other is **recessive**. Actually, it is the traits that are dominant or recessive rather than the alleles themselves; we refer to dominant and recessive alleles for the sake of convenience. Thus, one can say that the allele for type A blood is dominant to the type O allele. A heterozygous individual, with one A and one O allele, will have type A blood. The heterozygous (AO) individual will have exactly the same observed physical characteristic, or **phenotype**, as the homozygous (AA) individual, even though the two have a different genetic composition, or **genotype**. Only the homozygous recessive genotype (OO) will show the phenotype of type O blood.



➤ **Figure 2.11 Punnett Squares, Phenotype, and Genotype** These four Punnett squares (named for British geneticist Reginald Punnett) illustrate some of the possible phenotypes and genotypes of offspring within the A-B-O system. The alleles of one parent are listed on the left-hand side of the square, while the other parent's alleles are listed across the top. The potential genotypes of offspring are listed in the colored squares by letter. Offspring phenotypes are indicated by color: Blue indicates the type A phenotype; orange indicates the B phenotype. Individuals with one A and one B allele have the AB phenotype and make both blood antigens. Individuals with the O phenotype (magenta) have two O alleles.

The dominance of one allele does not mean that the recessive one is lost or in some way blended. A type A heterozygous parent (AO) will produce sex cells containing both A and O alleles. (This is an example of Mendel's law of segregation, that alleles retain their separate identities.) Recessive alleles can be handed down for generations before they show up in the phenotype when matched with another recessive allele in the process of sexual reproduction. The dominant allele masks the expression of the recessive allele.

Because all of the traits Mendel studied in garden peas showed this dominant–recessive relationship, it was believed for some years that this was the only relationship possible. Later studies show that, in some cases, neither allele is dominant; they are both *codominant.* Codominance in human heredity can be seen in the inheritance of blood types alleles A and B. Heterozygous individuals have a phenotype of AB because neither allele can dominate the other.

Blood types illustrate another complexity of heredity. Although we each have at most two alleles for any given gene—only one allele can appear on each of the two homologous chromosomes—the number of possible alleles for that gene found in a population is not limited to two. For example, over 100 alleles exist for **hemoglobin**, the blood protein that carries oxygen.

homozygous Refers to a chromosome pair that bears identical alleles for a single gene.

heterozygous Refers to a chromosome pair that bears different alleles for a single gene.

dominant In genetics, a term to describe the ability of an allele for a trait to mask the presence of another allele.

recessive In genetics, a term to describe an allele for a trait whose expression is masked by the presence of a dominant allele.

phenotype The observable characteristic of an organism that may or may not reflect a particular genotype due to the variable expression of dominant and recessive alleles.

genotype The alleles possessed for a particular trait.

hemoglobin The protein that carries oxygen in red blood cells.

polygenetic inheritance Two or more genes contributing to the phenotypic expression of a single characteristic.

Polygenetic Inheritance

So far, we have described traits of organisms that are determined by just one gene. However, multiple genes control most physical traits, such as body build, skin color, or susceptibility to disease. In such cases, we speak of **polygenetic inheritance**, in which the respective alleles of two or more genes influence phenotype. For example, several individuals may have the exact same height, but because there is no single height gene that determines an individual's size, it is impossible to neatly unravel the genetic underpinnings of 160 centimeters (5 ft. 3 in.). Characteristics subject to polygenetic inheritance exhibit a continuous range of variation in their phenotypic expression that does not correspond to simple Mendelian rules. As biological anthropologist Jonathan Marks demonstrates in the following Original Study, the relationship between genetics and continuous traits remains misunderstood.

ORIGINAL STUDY

Ninety-Eight Percent Alike: What Our Similarity to Apes Tells Us about Our Understanding of Genetics

By Jonathan Marks

It's not too hard to tell Jane Goodall from a chimpanzee. Goodall is the one with long legs and short arms, a prominent forehead, and whites in her eyes. She's the one with a significant amount of hair only on her head, not all over her body. She's the one who walks, talks, and wears clothing.

A few decades ago, however, the nascent field of molecular genetics recognized an apparent paradox: However easy it may be to tell Jane Goodall from a chimpanzee on the basis of physical characteristics, it is considerably harder to tell them apart according to their genes.

More recently, geneticists have been able to determine with precision that humans and chimpanzees are over 98 percent identical genetically, and that figure has become one of the most well-known factoids in popular scientific literature. It has been invoked to argue that we are simply a third kind of chimpanzee, together with the common chimp and the rarer bonobo; to claim human rights for nonhuman apes; and to explain the roots of male aggression.

Using the figure in those ways, however, ignores the context necessary to make sense of it. Actually, our amazing genetic similarity to chimpanzees is a scientific fact constructed from two rather more mundane facts: our familiarity with apes and our unfamiliarity with genetic comparisons.

To begin with, it is unfair to juxtapose differences between the bodies of people and apes with similarities in their genes. After all, we have been comparing bodies of humans and chimpanzees for 300 years, and we have been comparing DNA sequences for less than 20 years.

Now that we are familiar with chimpanzees, we quickly see how different they look from us. But when chimpanzees were a novelty, in the 18th century, scholars were struck by the overwhelming similarity of human and ape bodies. And why not? Bone for bone, muscle for muscle, organ for organ, the bodies of humans and apes differ only in subtle ways. And yet, it is impossible to say just how physically similar they are. Forty percent? Sixty percent? Ninety-eight percent? Three-dimensional beings that develop over their lifetimes don't lend themselves to a simple scale of similarity.

Genetics brings something different to the comparison. A DNA sequence is a one-dimensional entity, a long series of A, G, C, and T subunits. Align two sequences from different species and you can simply tabulate their similarities; if they match 98 out of 100 times, then the species are 98 percent genetically identical.

But is that more or less than their bodies match? We have no easy way to tell, for making sense of the question "How similar are humans and chimps?" requires a frame of reference. In other words, we should be asking: "How similar are humans and chimps, compared to what?" Let's try and answer the question. How similar are humans and chimps, compared to, say, sea urchins? Humans and chimpanzees have limbs, skeletons, bilateral symmetry, a central nervous system; each bone, muscle, and organ matches. For all intents and purposes, humans and chimpanzees aren't 98 percent identical, they're 100 percent identical.

On the other hand, when we compare the DNA of humans and chimps, what does the percentage of similarity mean? We conceptualize it on a linear scale, on which 100 percent is perfectly identical, and 0 percent is totally different. But the structure of DNA gives the scale a statistical idiosyncrasy.

Because DNA is a linear array of those four bases—A, G, C, and T—only four possibilities exist at any specific point in a DNA sequence. The laws of chance tell us that two random sequences from species that have no ancestry in common will match at about one in every four sites.

Moreover, the genetic comparison is misleading because it ignores qualitative differences among genomes. Genetic evolution involves much more than simply replacing one base with another. Thus, even among such close relatives as humans and chimpanzees, we find that a chimp's genome is estimated to be about 10 percent larger than a human's; that one human chromosome contains a fusion of two small chimpanzee chromosomes; and that the tips of each chimpanzee chromosome contain a DNA sequence that is not present in humans.

In other words, the pattern we encounter genetically is actually quite close to the pattern we encounter anatomically. In spite of the shock the figure of 98 percent may give us, humans are obviously identifiably different from, as well as very similar to, chimpanzees. The apparent paradox is simply a result of how mundane apes have become and how exotic DNA still is.

Adapted from Marks, J. (2000, May 12). 98% alike (What our similarity to apes tells us about our understanding of genetics). *The Chronicle of Higher Education*, B7. Copyright © 2000 by Chronicle of Higher Education. Reprinted with permission of the author.

Evolutionary Forces and Populations

At the level of an individual, genetic traits are transmitted from parent to offspring, enabling a prediction about the chances that any given individual will display some phenotypic characteristic. At the level of a group, genetics takes on additional significance. It reveals how evolutionary processes account for the diversity of life on earth.

A key concept in genetics is that of the **population**, or a group of individuals within which breeding takes place. **Gene pool** refers to all the genetic variants possessed by members of a population. Natural selection takes place within populations as some members contribute a disproportionate share of the next generation. Over generations, the relative proportions of alleles in a population change (biological evolution) according to the varying reproductive success of individuals within that population. In other words, at the level of population genetics, **evolution** can be defined as changes in allele frequencies in populations. This is also known as *microevolution*. Four evolutionary forces—mutation, genetic drift, gene flow, and natural selection—create and pattern biological diversity.

Mutation

Mutation, the ultimate source of evolutionary change, constantly introduces new genetic variation. Mutation occurs randomly. Although some mutations may be harmful or beneficial to individuals, most mutations are neutral. But in an evolutionary sense, random mutation is inherently positive: It provides the variation upon which the other evolutionary forces work. New body plans—such as walking on two legs compared to knuckle walking like our closest relatives, chimpanzees and gorillas—ultimately derived from a series of genetic mutations. A random mutation might create a new allele that modifies protein, making possible a novel biological task. Without the variation brought in through mutations, populations could not change over time in response to changing environments.

Mutations may arise whenever copying mistakes are made during cell division. This may involve a change in a single base of a DNA sequence or, at the other extreme, relocation of large segments of DNA, including entire chromosomes. As you read this page, the DNA in each cell of your body is being damaged (Culotta & Koshland, 1994). Fortunately, DNA repair enzymes constantly scan DNA for mistakes, slicing out damaged segments and patching up gaps. Moreover, for sexually reproducing species like humans, the only mutations of any evolutionary consequence are those occurring in sex cells because these cells form future generations.

New mutations arise continuously because no species has perfect DNA repair; thus all species continue to evolve. Environmental factors may increase the rate at which mutations occur (➤ **Figure 2.12**). These factors include certain dyes, antibiotics, and chemicals used in food preservation. Radiation, whether of industrial or solar origin, represents another important cause of mutations. Even stress can increase mutation rates (Chicurel, 2001). Ultimately, mutations confer versatility at the population level, making it possible for an evolving species to adapt more quickly to environmental changes. Remember, however, that mutations occur randomly and thus do not arise out of need for some new adaptation.

Genetic Drift

Genetic drift refers to chance fluctuations of allele frequencies in the gene pool of a population. Changes at the population level derive from random events at the individual level affecting that individual's survival. For example, an individual squirrel in good health and with a number of advantageous traits may be killed in a chance forest fire; a genetically well-adapted baby cougar may not live longer than a day if its mother gets caught in an avalanche, whereas weaker offspring of another cougar mother may survive.

In a large population, such accidents of nature are unimportant; the accidents that preserve individuals with certain alleles will be balanced out by the accidents that destroy them. However, in small populations, such averaging out may not be possible. Because today's global human population size is large, we might suppose that human beings are unaffected by genetic drift. But a chance event, like a rockslide that kills five people from a small town, say a population of 1,000, could significantly alter the frequencies of alleles in the local gene pool. Whenever biological variation is observed, whether in the distant past or present, it is always possible that chance events of genetic drift are responsible for it.

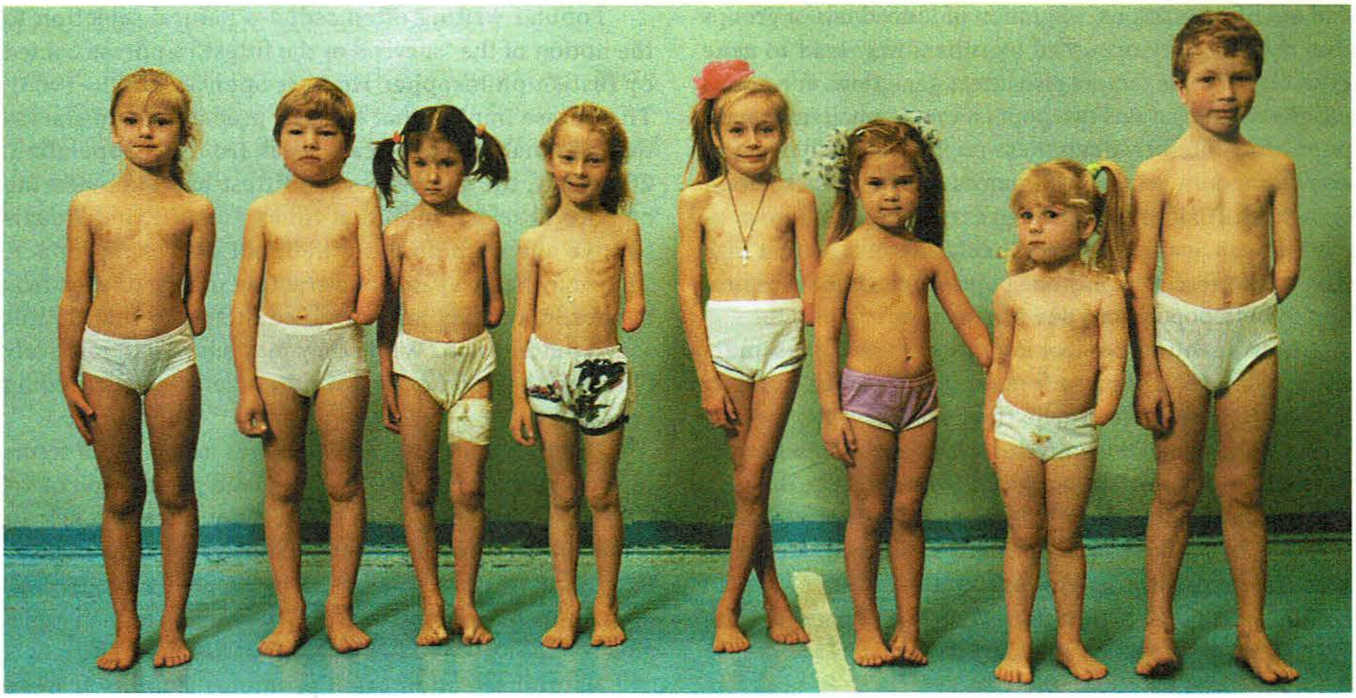
population In biology, a group of similar individuals that can and do interbreed.

gene pool All the genetic variants possessed by members of a population.

evolution The changes in allele frequencies in populations; also known as *microevolution*.

mutation The chance alteration of genetic material that produces new variation.

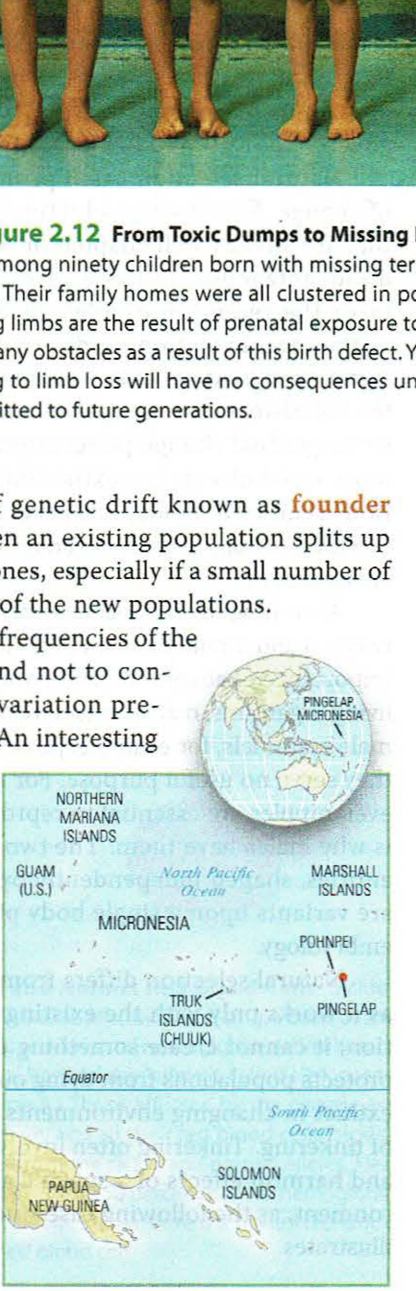
genetic drift The chance fluctuations of allele frequencies in the gene pool of a population.



© Gerg Ludwig/INSTITUTE

➤ **Figure 2.12 From Toxic Dumps to Missing Hands** These eight children, each missing a hand, were among ninety children born with missing terminal limbs over a twenty-year period in Moscow, Russia. Their family homes were all clustered in polluted industrialized sections of the city, and the missing limbs are the result of prenatal exposure to toxins. These children and their families certainly face many obstacles as a result of this birth defect. Yet from an evolutionary perspective, the mutations leading to limb loss will have no consequences unless they appear in the reproductive cells and are transmitted to future generations.

A specific kind of genetic drift known as **founder effects** may occur when an existing population splits up into two or more new ones, especially if a small number of individuals found one of the new populations. In such cases, the gene frequencies of the smaller population tend not to contain the full range of variation present in the larger one. An interesting example of founder effects can be seen on the Pacific Ocean island of Pingelap in Micronesia, where 5 percent of the population is completely color-blind, a condition known as *achromatopsia*. This is not the "normal" red–green color-blindness that affects 8 to 20 percent of males in most populations but rather a complete inability to see color. The high frequency of achromatopsia began around 1775, after a typhoon swept through the island, reducing its total population to only twenty individuals. Among the survivors was a single individual who was heterozygous for this condition. After a few generations, this gene became fully embedded in the expanding population. Today, a full 30 percent of the island's inhabitants are carriers of the color-blind gene, compared to a mere .003 percent seen in the United States (Sacks, 1998).



Gene Flow

Gene flow, or the introduction of new alleles from nearby populations, brings new genetic variation into a population: Interbreeding allows "road tested" genes to flow into

founder effects A particular form of genetic drift deriving from a small founding population not possessing all the alleles present in the original population.

gene flow The introduction of alleles from the gene pool of one population into that of another.

and out of populations. Migration of individuals or groups into the territory occupied by others may lead to gene flow. Geographic factors also affect gene flow. For example, if a river separates two populations of small mammals, preventing interbreeding, these populations will begin to accrue random genetic differences due to their isolation (genetic drift). If the river changes course and the two populations can again interbreed freely, new alleles that may have been present in only one population will now be present in both populations due to gene flow.

Among humans, social factors—such as mating rules, intergroup conflict, and our ability to travel great distances—affect gene flow. For example, the last 500 years have seen the introduction of alleles into Central and South American populations from both the Spanish colonists and the Africans whom Europeans imported as slaves. More recent migrations of people from East Asia have added to this mix. Throughout the history of human life on earth, gene flow has prevented populations from developing into separate species.

Natural Selection

Although gene flow and genetic drift may produce changes in the allele frequency of a population, that change would not necessarily make the population better adapted to its biological and social environment. Natural selection, the evolutionary force described by Darwin, accounts for adaptive change. **Adaptation**—a series of beneficial adjustments to a particular environment—is the outcome of natural selection. As we will explore throughout this text, humans can adapt to their environment through culture as well as biology. When biological adaptation occurs at a genetic level, natural selection is at work.

Natural selection shapes genetic variation at the population level to fit local environmental conditions. In other words, instead of random individuals passing their traits on to the next generation, selection by the forces of nature favors some individuals over others. In the process, the frequency of genetic variants for harmful or nonadaptive traits within the population reduces while the frequency of genetic variants for adaptive traits increases. Over time, changes in the genetic structure of the population can result in formation of new species.

Popular writing often reduces natural selection to the notion of the "survival of the fittest," a phrase coined by British philosopher Herbert Spencer (1820–1903). The phrase implies that disease, predation, and starvation eliminate the physically weak from the population. Obviously, the survival of the fittest has some bearing on natural selection. But at times, "less fit" individuals survive and even do quite well, but do not reproduce. They may be incapable of attracting mates, or they may be sterile, or they may produce offspring that do not survive after birth. While they may survive to relatively old ages, they do not pass on their genes to succeeding generations.

Ultimately, all natural selection is measured in terms of **reproductive success**—mating and production of viable offspring who will in turn carry on one's genes. In some human societies, a woman's social worth is assessed in terms of reproductive success or her ability to bear children. In these contexts, infertility becomes a human rights issue.

Natural selection often promotes stability instead of change. **Stabilizing selection** occurs in populations that are already well adapted or where change would be disadvantageous. In these cases, natural selection will favor the retention of allele frequencies more or less as they are. Evolution tends not to proceed as a steady, stately progression over vast periods of time. Instead, the life history of most species consists of relative stability or gradual change punctuated by shorter periods of more rapid change (or extinction) when altered conditions require new adaptations or when a new mutation produces an opportunity to adapt to some other available environment.

According to the fossil record, most species survive between 3 and 5 million years (Thomson, 1997). Despite the importance of adaptation and natural selection in shaping living organisms, many traits have no adaptive function. All male mammals, for example, possess nipples, even though they serve no useful purpose. For female mammals, however, nipples are essential to reproductive success, which is why males have them. The two sexes are not separate entities, shaped independently by natural selection, but are variants upon a single body plan, elaborated in later embryology.

Natural selection differs from the concept of design as it works only with the existing store of genetic variation; it cannot create something entirely new. Variation protects populations from dying out or species from going extinct in changing environments. Evolution is a process of tinkering. Tinkering often involves balancing beneficial and harmful effects of a specific allele in a specific environment, as the following case study of sickle-cell anemia illustrates.

adaptation A series of beneficial adjustments to a particular environment.

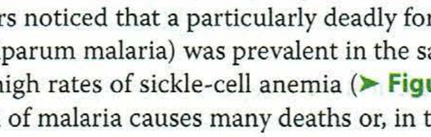
reproductive success The relative production of fertile offspring by a genotype. In practical terms, the number of offspring produced by individual members of a population is tallied and compared to that of others.

stabilizing selection Natural selection acting to promote stability rather than change in a population's gene pool.

The Case of Sickle-Cell Anemia

Among human beings, **sickle-cell anemia** is a particularly well-studied case of adaptation. This painful disease, in which oxygen-carrying red blood cells change shape (sickle; ➤ **Figure 2.13**) and clog the finest parts of the circulatory system, first came to the attention of geneticists in Chicago who observed that the disease disproportionately impacted African Americans. Further investigation found that populations that live in a clearly defined belt across Central Africa had the sickle-cell allele at surprisingly high frequencies. Geneticists were curious to know why such a harmful hereditary disability persisted in these populations. According to the theory of natural selection, any alleles that are harmful tend to disappear from a group because individuals who are homozygous for the abnormality generally die—are "selected out"—before they reproduce. Why, then, has this seemingly harmful condition persisted in populations from Central Africa?

The answer to this mystery began to emerge when researchers noticed that a particularly deadly form of malaria (falciparum malaria) was prevalent in the same areas that had high rates of sickle-cell anemia (➤ **Figure 2.14**). This form of malaria causes many deaths or, in those who survive, high fevers that interfere with individuals' reproductive abilities. Moreover, researchers discovered hemoglobin abnormalities among people living in parts of the Arabian Peninsula, Greece, Algeria, Syria, and India, all regions where malaria is (or was) common. Thus, selection favored heterozygous individuals with normal and sickling hemoglobin. The loss of alleles for abnormal hemoglobin caused by the death from sickle-cell anemia was balanced out by the loss of alleles for normal hemoglobin, as those homozygous for normal hemoglobin were more likely to die from malaria.



➤ **Figure 2.13 Sickle and Normal Red Blood Cells** Sickle-cell anemia is caused by a genetic mutation in a single base of the hemoglobin gene, resulting in abnormal hemoglobin, called hemoglobin S or Hb^{S}. (The normal hemoglobin allele is called Hb^{A}, not to be confused with blood type A.) Those afflicted by the disease are homozygous for the Hb^{S} allele, and all their red blood cells "sickle." Codominance is observable with the sickle and normal alleles. Heterozygotes (genotype $\text{Hb}^{A}\,\text{Hb}^{S}$) make 50 percent normal hemoglobin and 50 percent sickle hemoglobin. Shown here is a sickled red blood cell compared to a normal red blood cell.

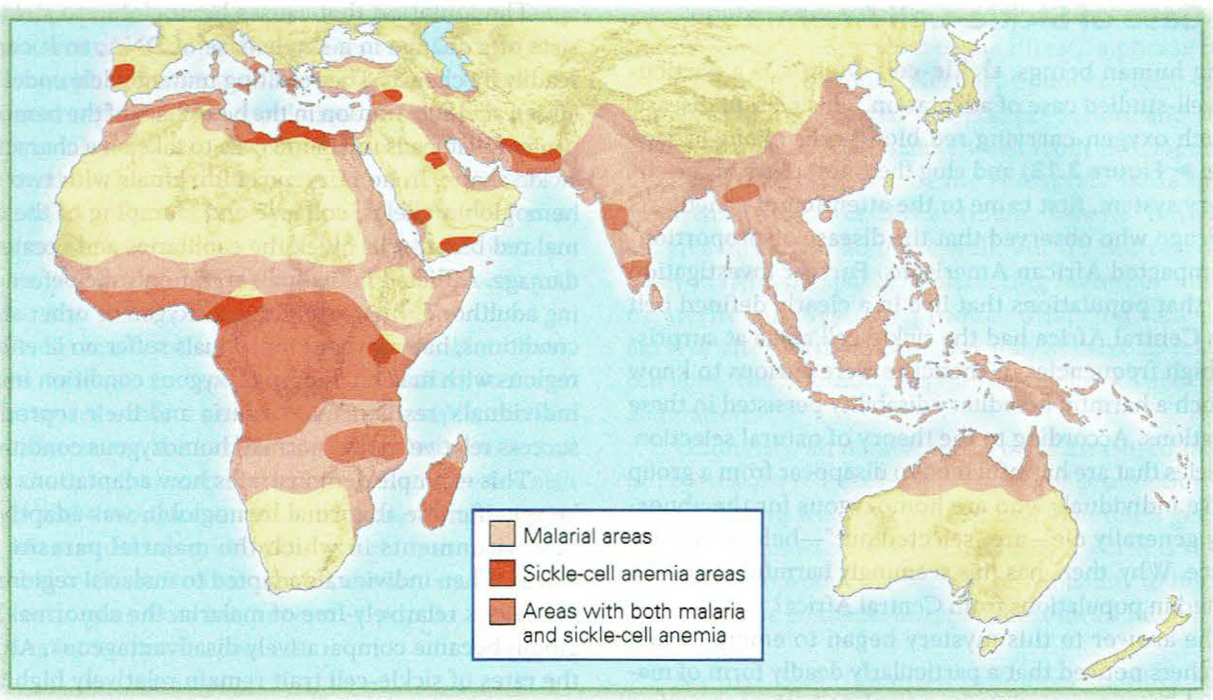
The mutation that causes hemoglobin to sickle consists of a change in a single base of DNA, so it can arise readily by chance. The resulting mutant allele codes for an amino acid substitution in the beta chain of the hemoglobin protein that leads red blood cells to take on a characteristic sickle shape. In homozygous individuals with two sickle-hemoglobin alleles, collapse and clumping of the abnormal red blood cells block the capillaries and create tissue damage. Afflicted individuals commonly die before reaching adulthood. Except under low oxygen or other stressful conditions, heterozygous individuals suffer no ill effects. In regions with malaria, the heterozygous condition improves individuals' resilience to malaria and their reproductive success relative to the "normal" homozygous condition.

This example demonstrates how adaptations tend to be specific; the abnormal hemoglobin was adaptive only in environments in which the malarial parasite flourished. When individuals adapted to malarial regions came to regions relatively free of malaria, the abnormal hemoglobin became comparatively disadvantageous. Although the rates of sickle-cell trait remain relatively high among African Americans—about 9 percent have the sickling trait—this has significantly declined from the 22 percent estimated among the first African captives who were sold as slaves. A similar decline in the sickle-cell allele would occur over the course of several generations in malarial zones if this deadly disease were brought under control.

This example also illustrates the role culture plays in biological adaptation. In Africa, the severe form of malaria was not a significant problem until humans abandoned food foraging for farming a few thousand years ago. To farm, people had to clear areas of the natural forest cover. In the forest, decaying vegetation on the forest floor gave the ground an absorbent quality so that the heavy rainfall rapidly soaked into soil. But once stripped of its natural vegetation, the soil lost this quality. In addition, without the forest canopy to break the force of rainfall, heavy rains compacted the soil further. The stagnant puddles that subsequently formed provided the perfect breeding environment for the type of mosquito that hosts the malarial parasite. These mosquitoes then began to flourish and transmit the malarial parasite to humans.

Thus, humans unwittingly created the kind of environment that made a disadvantageous trait—the abnormal hemoglobin associated with sickle-cell anemia—advantageous. Although the biological process of evolution accounts for frequency of the sickle-cell allele, cultural processes shape the environment to which humans adapt.

sickle-cell anemia An inherited form of anemia produced by a mutation in the hemoglobin protein that causes the red blood cells to assume a sickle shape.



➤ **Figure 2.14 The Distribution of Malaria and the Sickle-Cell Allele** In regions with a high incidence of falciparum malaria, people native to these areas have a higher than normal rate of the allele that causes sickle-cell anemia. Researchers have surmised that natural selection preserved the allele for the sickle-cell trait to protect individuals from the devastating effects of malaria.

Adaptation and Physical Variation

Anthropologists study biological diversity in terms of **clines**, or the continuous gradation over space in the form or frequency of a trait. The spatial distribution or cline for the sickle-cell allele allowed anthropologists to identify the adaptive function of this gene in a malarial environment. Clinal analysis of a continuous trait such as body shape, which is controlled by a series of genes, allows anthropologists to interpret human global variation in body build as an adaptation to climate.

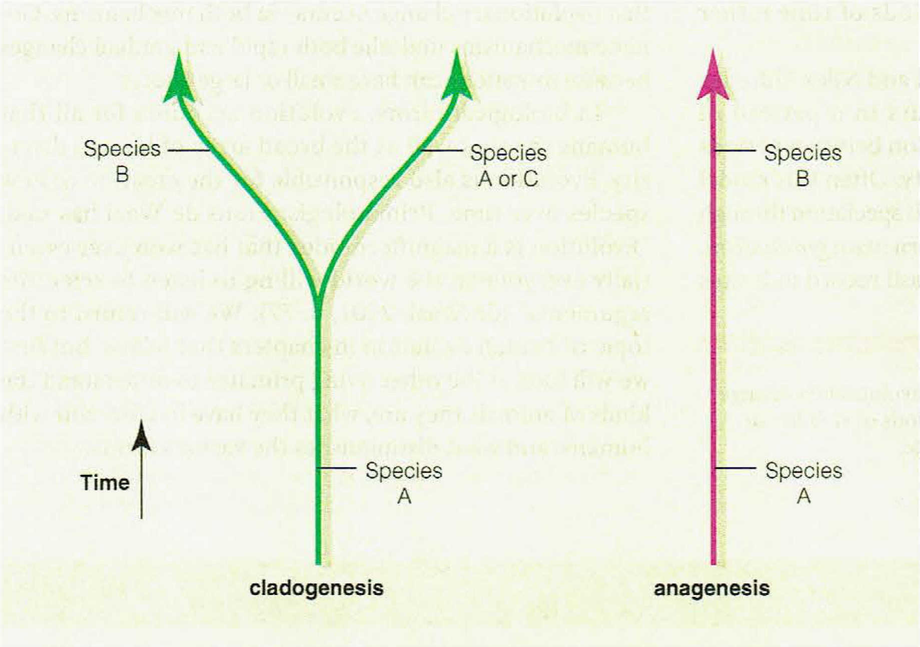
Generally, people long native to regions with cold climates tend to have greater body bulk (not to be equated with fat) relative to their extremities (arms and legs) than do people native to regions with hot climates, who tend to be relatively tall and slender. In hot, open country people benefit from a long, slender body that can get rid of excess heat quickly. A small slender body can also promote heat loss due to a high surface area to volume ratio. A person with larger body bulk and relatively short extremities may suffer more from summer heat, but this person will conserve needed body heat under cold conditions because a bulky body has less surface area relative to volume.

Climate can also contribute to human variation through its impact on the process of growth and development (developmental adaptation). For example, some of the biological mechanisms for withstanding cold or dissipating heat have been shown to vary depending upon the climate individuals experience as children. People spending their youth in very cold climates develop circulatory system modifications that allow them to remain comfortable at temperatures that those from warmer climates cannot tolerate. Similarly, hot climate promotes development of a higher density of sweat glands, creating a more efficient system for sweating to keep the body cool.

Cultural processes complicate studies of biological adaptation to climate. For example, a poor diet during childhood affects the growth process and ultimately impacts adult body shape and size. Clothing also complicates these studies. In fact, culture rather than biology accounts for much of the way people adapt to cold. For instance, to cope with bitter Arctic climates, the Inuit peoples of northern Canada (and other Eskimos) created artificial tropical environments for themselves inside their clothing. Such cultural adaptations allow humans to inhabit the entire globe.

---

clines The gradual changes in the frequency of an allele or trait over space.



➤ **Figure 2.15 Mechanisms of Speciation** Cladogenesis occurs as different populations of an ancestral species become reproductively isolated. Through genetic drift and differential selection, the number of descendant species increases. Speciation by anagenesis can take place as small differences in traits that (by chance) are advantageous in a particular environment accumulate in a species' gene pool, eventually transforming an old species into a new one. Genetic drift may also account for anagenesis.

Macroevolution and the Process of Speciation

While *microevolution* refers to changes in the allele frequencies of populations, **macroevolution** focuses on **speciation**—the formation of new species—and on the evolutionary relationships among groups of species. The microevolutionary forces of mutation, genetic drift, gene flow, and natural selection can lead to macroevolutionary change as species diverge.

As defined earlier in the chapter, *species*—a population or group of populations capable of interbreeding and producing viable, fertile offspring—are reproductively isolated. The bullfrogs in one farmer's pond are the same species as those in a neighboring pond, even though the two populations may never actually interbreed; in theory, they could interbreed if brought together. But isolated populations may be in the process of evolving into different species, and it is hard to tell exactly when they become distinct.

Factors, known as *isolating mechanisms*, can separate breeding populations and lead to the appearance of new species. Because isolation prevents gene flow, changes that affect the gene pool of one population cannot be introduced into the gene pool of another. Random mutation may introduce new alleles in one of the isolated populations but not in the other. Genetic drift and natural selection may affect the two populations in different ways. Over time, as the two populations continue to differ from each other, speciation occurs in a branching fashion known as **cladogenesis**. Speciation can also happen without branching, as a single population accumulates sufficient new mutations over time to be considered a separate species. This process known as **anagenesis** (➤ **Figure 2.15**) appears in the fossil record when a group of organisms takes on a different appearance over time.

Speciation can occur at various rates. Scholars generally consider speciation through natural selection to occur at a slow rate as organisms become better adapted to their environments. Sometimes, however, speciation can occur quite rapidly. For example, a genetic mutation such as one involving a key *regulatory gene,* a gene that turns other genes off and on, can lead to formation of a new body plan. Such genetic accidents may involve material that is broken off, transposed, or transferred from one chromosome to another.

Genes that regulate the growth and development of an organism may have a major effect on its adult form. Scientists have discovered certain key genes called *homeobox genes* that are responsible for large-scale effects on the growth and development of an organism. If a new body plan happens to be adaptive, natural selection will

macroevolution	Evolution above the species level or leading to the formation of new species.
speciation	The process of forming new species.
cladogenesis	Speciation through a branching mechanism whereby an ancestral population gives rise to two or more descendant populations.
anagenesis	A sustained directional shift in a population's average characteristics.

maintain this new form for long periods of time rather than promoting change.

Paleontologists Stephen Jay Gould and Niles Eldredge have proposed that speciation occurs in a pattern of **punctuated equilibria**, or the alternation between periods of rapid speciation and times of stability. Often this model of evolutionary change is contrasted with speciation through adaptation, sometimes referred to as *Darwinian gradualism*. A close look at the genetics and the fossil record indicates that evolutionary change occurs via both mechanisms. Genetic mechanisms underlie both rapid and gradual changes because mutations can have small or large effects.

punctuated equilibria A model of macroevolutionary change that suggests evolution occurs via long periods of stability or stasis punctuated by periods of rapid change.

In biological terms, evolution accounts for all that humans share as well as the broad array of human diversity. Evolution is also responsible for the creation of new species over time. Primatologist Frans de Waal has said, "Evolution is a magnificent idea that has won over essentially everyone in the world willing to listen to scientific arguments" (de Waal, 2001, p. 77). We will return to the topic of human evolution in chapters that follow, but first we will look at the other living primates to understand the kinds of animals they are, what they have in common with humans, and what distinguishes the various forms.

Chapter Checklist

How does evolutionary theory differ from creation stories?

✓ Scientific theories are based on testable hypotheses.

✓ Scientific theories provide mechanisms to account for the diversity of life on earth.

How are living things classified, and how did this system come about?

✓ The science of taxonomy classifies living organisms into a series of progressively more inclusive categories on the basis of visual similarities and developmental patterns.

✓ Carolus Linnaeus devised the *Systema Naturae*, the first system to classify living things on the basis of similarities in body structure, body function, and sequence of bodily growth.

✓ Modern taxonomy adds genetic characteristics to the basic Linnaean system.

✓ Species, the smallest working units in biological classificatory systems, are reproductively isolated populations or groups of populations capable of interbreeding to produce fertile offspring.

What is evolution, and when was this central biological theory formulated?

✓ Charles Darwin formulated his theory of evolution in 1859, based on differential reproductive success among members of a population that become adapted to their environment through natural selection.

✓ Four evolutionary forces—mutation, genetic drift, gene flow, and natural selection—affect the genetic structures of populations. Evolution at the level of population genetics (also known as microevolution) is change in allele frequencies.

✓ Macroevolution focuses on the formation of new species (speciation) and on the evolutionary relationships among groups.

What is the molecular basis of evolution?

✓ Genes, the units of heredity, are segments of molecules of DNA (deoxyribonucleic acid). The entire sequence of DNA is known as the genome.

✓ DNA resembles two strands of rope twisted around each other with ladderlike rungs connecting the two strands.

✓ The sequence of bases along the DNA molecule directs the production of proteins. Proteins constitute specific identifiable traits such as blood type.

✓ Different versions or alternate forms of a gene for a given trait are called alleles. The total number of different alleles of genes in a population is its gene pool.

How do cells and organisms reproduce?

✓ DNA molecules can replicate or produce exact copies of themselves so that new daughter cells will be exact genetic copies of the parent cell.

✓ DNA molecules are located on chromosomes, structures found in the nucleus of each cell. Chromosomes consist of two sister chromatids.

✓ Each species has a characteristic number of chromosomes, found in pairs in sexually reproducing organisms. Humans have twenty-three pairs of chromosomes.

✓ Mitosis is the kind of cell division that results in two identical daughter cells.

✓ Meiosis results in four daughter cells each containing half the number of chromosomes of the parent cell. Fertilization, the union of an egg and a sperm cell, reestablishes the normal number.

How do different traits get inherited across generations?

✓ Individuals inherit traits independently from each parent.

✓ Dominant alleles are able to mask the presence of recessive alleles. *Codominant* alleles are both expressed when present.

- *Phenotype* refers to the physical characteristics of an organism. *Genotype* refers to its genetic composition. Two organisms may have different genotypes but the same phenotype.

How do the four evolutionary forces contribute to the diversity of life on earth?

- Mutation provides the ultimate source of genetic variation.
- *Genetic drift* refers to the effects of random events on the gene pools of populations.
- Gene flow, the introduction of new variants of genes from nearby populations, distributes new variation to all populations and prevents speciation.
- Natural selection reduces the frequency of alleles for harmful or maladaptive traits within a population and increases the frequency of alleles for adaptive traits.

What are some examples of human adaptation through natural selection?

- The sickle-cell trait, caused by the inheritance of an abnormal form of hemoglobin, is an adaptation to life in regions in which malaria is common.
- People native to cold climates tend to have greater body bulk relative to their extremities while individuals from hot climates tend to be relatively tall and slender.

How are new species formed?

- Speciation can occur in a branching fashion (cladogenesis) or without branching (anagenesis) as a population accumulates sufficient new mutations over time to be considered a separate species.
- Microevolutionary forces can lead to macroevolutionary change, but the tempo of evolutionary change varies.
- Mutations in regulatory genes can bring about rapid change. According to the punctuated equilibrium model, macroevolution is characterized by long spans of relative stability interspersed with periods of rapid change.

Questions for Reflection

1. Have genetics and DNA become a part of your everyday experience? If so, how? Has genetics challenged your conception of what it means to be human? How much of your life, or of the lives of people around you, is dictated by the structure of DNA?
2. Scientific fact and theory can challenge other belief systems. Is it possible for scientific models of human evolution and religious stories of creation to coexist? How do you personally reconcile science and religion?
3. The four evolutionary forces—mutation, genetic drift, gene flow, and natural selection—all affect biological variation. Some are at work in individuals while others function at the population level. Compare and contrast these evolutionary forces, outlining their contributions to biological variation.
4. The frequency of the sickle-cell allele in populations provides a classic example of adaptation on a genetic level. Describe the benefits of this deadly allele. Are mutations good or bad?

DIGGING INTO ANTHROPOLOGY

Making Meaning of Memes

Memes going viral on the Internet might seem distant from biology and genetics. But they share a common ancestry. Evolutionary biologist Richard Dawkins coined the term *meme* to refer to an abstract unit of cultural transmission, analogous to the role of the gene in biology. Arguing that cultural information is subject to many of the same evolutionary forces as biological attributes, Dawkins's idea gave rise to the field of memetics, drawing in scholars from a variety of disciplines. A meme can be virtually anything that self-replicates (spreads from person to person), including phrases, songs, gestures, fashion trends, viral videos, rituals, religious traditions, cuisine, technologies, and so on. Choose a contemporary meme and investigate its hereditary history, considering how the evolutionary forces discussed in this chapter have played into its cultural pathway. When and how has it mutated? What other meme "alleles" has it competed with for reproductive success? How has it adapted to different cultural environments? On the other hand, how does the meme fail to conform with genetic hereditary processes?